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Brown et al.

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## [54] METHODS AND COMPOSITIONS FOR THE DETECTION OF FAMILIAL HYPERCHOLESTEROLEMIA

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[\*] Notice: The portion of the term of this patent subsequent to May 17, 2005 has been disclaimed.

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[58] Field of Search ..... 435/6, 803, 91; 436/501, 811; 935/78, 9, 19, 77; 530/808, 825

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### [57]

### ABSTRACT

Recombinant DNA transfer vectors containing DNA inserts which are complementary to either the human LDL receptor gene, or its mRNA transcript, are disclosed. Also disclosed are methods which utilize these genetic probes for diagnosing Familial Hypercholesterolemia (FH) in a suspected individual. A case study of numerous such individual are disclosed wherein the genetic deletion mutation is detailed with great precision through the practice of this invention.

26 Claims, 16 Drawing Sheets